Primary Autoimmune Cerebellar Ataxia (PACA)

Information for patients
Neurology
What is ataxia?

Ataxia is the name given to a group of neurological symptoms and signs that affect balance, coordination, speech and sometimes vision. There are different causes of ataxia and people are affected in different ways. Ataxia usually results from damage to the balance organ of the nervous system called the cerebellum, which is situated at the back of the brain.

What is autoimmunity?

When your own immune system reacts inappropriately to a trigger (this could be an infection or other environmental factors, some of which are not known) it may result in damage to different parts of your own body. We refer to this group of diseases as autoimmune diseases. One of the most common autoimmune diseases is insulin dependent diabetes (sometimes referred to as type 1 diabetes mellitus) where the damage is to the pancreas causing inability to produce insulin. Another example is underactive thyroid disease (damage to the thyroid gland and inability to produce thyroxine).

What is Primary Autoimmune Cerebellar Ataxia (PACA)?

The balance organ of the nervous system (cerebellum) can also be a target of autoimmunity. Examples of autoimmune ataxia where the trigger is known include gluten ataxia (a result of eating gluten protein found in wheat, barley and rye) or post-infectious cerebellitis (ataxia following an infection).

In the majority of cases of autoimmune ataxia there is no obvious trigger. When this happens we refer to this condition as primary autoimmune cerebellar ataxia (PACA). Whilst there is no specific marker (e.g. blood tests) for PACA there are several clues that ataxia specialists
look for in order to identify PACA, for example the patient with suspected PACA may already have another autoimmune disease (e.g. underactive thyroid) or have a family history of autoimmune diseases. Despite such clues, all patients will have had extensive investigations to rule out other common causes of ataxia (including genetic testing for genetic causes) before the final diagnosis of PACA is reached.

Is there a treatment?

In some autoimmune diseases that result in permanent damage of a particular body organ, the treatment can be simple, for example in autoimmune hypothyroidism, taking thyroxine resolves the symptoms. In type 1 diabetes, insulin is the treatment. Unfortunately in the case of cerebellar ataxia due to PACA there is no medication that can repair the damage and brain cells have no capacity for regenerating, so the aim would be to stop the progression of the disease. To do this we sometimes use drugs that suppress the immune system (also known as immunosuppressants). No large-scale treatment trials have been done to assess the use of such drugs in PACA. However there are case reports and small series of patients with ataxia that benefit from the use of such drugs. The Sheffield Ataxia Centre has extensive experience with the treatment of PACA.

I have been diagnosed with PACA – what happens now?

The decision to treat with drugs that suppress the immune system will be based on whether the ataxia appears to be progressive. This can be assessed both by clinical examination (sometimes we use a monitoring scale for assessing the severity of the ataxia) but also with the use of brain imaging called Magnetic Resonance Spectroscopy of the cerebellum. This technique has been in use at the Sheffield Ataxia
Centre since 2008 and is a very accurate way of assessing how well the nerve cells function within the balance centre and a reliable method for monitoring the disease.

Your Consultant will discuss with you the suspected diagnosis of PACA and the possibility of using immunosuppressive drugs to stop the progress of the disease. A joint decision will be made about the treatment and your GP will be approached to ensure that they can help with the blood monitoring.

Immunosuppressant drugs require regular safety blood monitoring, at first on a very frequent basis (every 2 weeks). Eventually the monitoring is done every few months. An MR spectroscopy scan will be done before you start the medication so that it can be used as a baseline measurement for monitoring purposes. The medication will then be prescribed with clear instructions of how to take it and what monitoring arrangements are in place. Adjustments to the dose of the medication are usually done during the first follow up appointment (usually 1 month after starting the medication). Blood testing is done when you attend the ataxia clinic and also at the GP’s practice, usually by the district nurse.

Information about the specific immunosuppressant drug that is likely to be used will be provided separately.

**How will the effect of the drug be monitored?**

You will be seen at regular intervals both by the Sheffield Ataxia Centre Neurology Consultant and the monitoring nurse, as well as the Ataxia Nurses. After about a year on the medication you may have a repeat MR spectroscopy brain scan to monitor if there has been an improvement by comparison to the baseline scan. Assessment of the ataxia will also be done by repeating the ataxia rating scale done before starting the medication.
Will my ataxia get better?

The aim of the treatment is to prevent further damage to the balance centre, stabilise the disease process and prevent progression of the ataxia. Some people find that their symptoms improve, whilst for others this is not quite so obvious. This sometimes depends on the severity of the ataxia. We also rely on you to tell us how you feel things are going in terms of stabilisation or improvement. Additional information will be obtained from the brain scans and the ataxia rating scale.

Where can I find out more about ataxia?

Ataxia UK

Ataxia UK is the national charity for everyone affected by ataxia. Membership is free. In addition to offering a range of support services, they fund research into treatments, organise conferences and produce a newsletter to keep you updated about all issues to do with ataxia. For more information you can contact Ataxia UK as follows:

- 0845 644 0606 (Helpline)
- 0207 582 1444
- www.ataxia.org.uk